A simple sequence repeat polymorphism at the human growth hormone locus

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Source/Description: The polymorphic simple sequence repeat begins at the 25765 base pair of the human growth hormone locus on chromosome 17q22-24 (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (2). The predicted length of the amplified sequence was 243 bp.

Primer Sequences: TCCAGCCTCGGAGACAGAAT (AAAG strand); AGTCCTTTCTCCAGAGCAGGT (CTTT strand).

Frequency: Estimated from 44 chromosomes of unrelated individuals. Observed heterozygosity = 82.6%.

Allele (bp)	Frequency	Allele (bp)	Frequency
253	0.02	221	0.14
251	0.02	219	0.07
247	0.02	217	0.20
244	0.04	215	0.07
241	0.04	213	0.04
239	0.04	211	0.04
233	0.04	209	0.04
231	0.04	205	0.04
229	0.02	201	0.02

Mendelian Inheritance: Co-dominant segregation was observed in two informative families.

Chromosomal Localization: The human growth hormone locus has been assigned to chromosome 17q22-24 (1).

Other Comments: The PCR reaction was performed on 80 ng of genomic DNA using 100 pmoles of each oligonucleotide primer. The samples were processed as described (3) except that the denaturation cycle at 94°C was extended to 1.4 minutes. The dinucleotide repeat was based on a (AAAG)₃ACA(AG)₃(AAA- $G_{11}(AG)_{11}(AAAG)_{17}$ sequence.

References: 1) Chen, E.Y. et al. (1989) Genomics 4, 479-497. 2) Weber, J.L. and May, P.E. (1989) Am. J. Hum. Genet. 44, 388-396. 3) Weber, J.L. et al. (1990) Nucl. Acids Res. 18, 4637.

Dinucleotide repeat polymorphism at the MAOA locus

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Source/Description: A human genomic clone selected by hybridization to a monoamine oxidase-A (MAOA) cDNA (Hsu et al. 1988) was subcloned into mp19. Hybridization with poly(dC- dA)·poly(dG-dT) allowed selection of a clone designated MAOCA-1. Sequencing of this clone provided the information necessary for primer synthesis. The predicted length of the amplified fragment was 114 bp.

Primer Sequences: AGAGACTAGACAAGTTGCAC (CA strand) CACTATCTTGTTAGCTCACT (GT strand)

Frequency: Estimated from 86 chromosomes from unrelated Caucasians. PIC = 0.69

Allele (bp)	Frequency	
C1 126	0.023	
C2 122	0.035	
C3 120	0.221	
C4 118	0.023	
C5 116	0.209	
C6 114	0.430	
C7 112	0.058	

Chromosomal Localisation: MAO-A has been assigned to Xp11.3 by in situ hybridization (Levy et al. 1989).

Mendelian Inheritance: Co-dominant X-linked inheritance was observed in 6 three generation and 4 two generation families.

Other Comments: Conditions of the PCR amplification are as follows: 33 cycles of 30 seconds @ 94°, 45 seconds @ 56°, and 45 seconds @ 70°. The final elongation cycle was 4 minutes @ 72°. Sizes of the alleles were compared with pUC9 cut with HpaII. The dinucleotide sequence was of the form $(AC)_{18}CG(AC)_3$.

The sequence of MAOCA-1 has been submitted to EMBL, accession no. X55451.

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References: 1) Hsu, Y.-P.P., Weyler, W., Chen, S., Sims, K.B., Rinehart, W.B., Utterback, M.C., Powell, J.F. Breakefield, X.O. (1988) J. Neurochem. 51, 1321-1324. 2) Levy, E.R., Powell, J.F., Buckle, V.J., Hsu, Y.-P.P., Breakefield, X.O. and Craig, I.W. (1989) Genomics 4, 368-370.

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